Forensic autopsy in sudden cardiac death

Autopsia forense en la muerte súbita cardíaca

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Received: April 18, 2017
Accepted: April 18, 2017

Key words: Sudden cardiac death, Autopsy, Forensic Medicine
Palabras clave: Muerte súbita cardíaca, Autopsia, Medicina Legal

To the Editor:

The sudden cardiac death represents an epidemiological problem of great social impact with an estimated incidence of 4000 cases/year, in apparently healthy individuals (16-60 years of age) in Spain.

This type of death is usually judicialized, given the unexpectedness of its occurrence and the absence of any background that justifies it and, therefore, the medical-legal autopsy has to be carried out, a situation that is desirable as recommended by the European regulations (Council of Ministers of the Member States of the European Union, Recommendation No. 99 on methodological harmonization of medical-legal autopsies).

At the Institute of Legal Medicine and Forensic Sciences of Valencia are performed, among others, autopsies of sudden and unexpected deaths in children under a year of age, and deceased up to 55 years, of presumably cardiovascular cause. In the figure are shown some statistical data. La muestra algunos datos estadísticos.

These autopsies are performed by the physicians assigned to the Pathology Department, following the protocols developed for these cases.

AUTOPSY IN SUDDEN CARDIAC DEATH

The case of a sudden death should be brought to the attention of the judicial authority, who will order to proceed with the removal of the body, either by the judicial commission or delegating to the forensic doctor (Code of Criminal Procedure, art. 7781), and the completion of the medical-legal autopsy.

In a case of sudden death, the objective of the autopsy will be2,3:
- To determine if the death was due to a cardiovascular disease.
- If it is a sudden cardiovascular death, to typify it and try to establish whether the mechanism was arrhythmic or mechanical.
- If the cardiovascular disease of the sudden death can be hereditary.
- To assess the possibility of drug abuse.

In the Institute of Forensic Medicine and Forensic Sciences of Valencia has been established a protocol for action in cases of sudden death of cardiac origin, whose purpose is:
- To identify the cause of death: hypertrophic, dilated, arrhythmogenic cardiomyopathy.
- To provide morphological data of interest that will have an impact on clinical assessment.
- Collection of biological samples for toxicological and genetic studies.
- To conduct the family members to multidisciplinary work groups.

According to the results of the autopsies, sudden cardiac deaths are classified in the following groups:
1. Structurally normal heart (where channelopathies are suspected).
2. Cardiomyopathies (whether hypertrophic, dilated, non-compactcd, arrhythmogenic, or not de-
3. Dissection of non-atheromatous thoracic aorta (it includes bicuspid aorta).

PROTOCOLS

The protocol of lifting the body includes important data relating to the circumstances of death (activity in the last hours, prior symptoms that indicate the time interval from the onset of symptoms, medical care received, among others) pathological background of the deceased (it is essential to know if there is a history of sudden death in the family); as well as getting a family member's contact phone number. These data will be later corroborated and expanded in the interview with a family member.

The autopsy protocol systematizes, among other data, the collection of anthropometric data (size, weight, perimeters) and the collection of samples:
- **Blood:** for genetic, lipid profile and toxicology studies. In the latter, a determination of the main drug abuse is requested: opiates, cocaine, cannabinoids, ethyl alcohol, among others. Blood is essential for the cardiogenic study.
- **Urine:** for the toxicological study and if it is a sudden death associated with sports. The presence of leukocytes, nitrites, urobilinogen, proteins, pH, blood, density, ketone bodies, bilirubin and glucose are also performed on a test strip.
- **Vitreous humor:** for determination of alcohol.
- **Hair:** for toxicological study, if applicable.
- **Tissue samples:** myocardial, liver and spleen for genetic studies.
- **Aorta:** to take a sample of the artery in the aortic dissections.
- **Histopathology:** there are obtained samples of the thyroids, rib, of the five pulmonary lobes, heart with 4 cm of the aortic and pulmonary trunks, of right lobe of liver, spleen, kidney and adrenal glands.

**Figure.** Statistical data of the Valencia’s forensic subunit.
A clinico-pathological update in arrhythmogenic right ventricular cardiomyopathy

Actualización clínico-patológica en la miocardiopatía arritmogénica del ventrículo derecho

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To the Editor:

Arrhythmogenic cardiomyopathy (AC) is a heart muscle disease characterized clinically by life-threatening ventricular arrhythmias and pathologically by an acquired and progressive dystrophy of the ventricular myocardium with fibro-fatty replacement. With an estimated prevalence of 1:2000-1:5000, AC is listed among rare diseases. A familial background consistent with an autosomal-dominant trait of inheritance is present in most of AC patients; recessive variants have also been reported, either or not associated with palmoplantar keratoderma and woolly hair. AC-causing genes mostly encode major components of the cardiac desmosomes and up to 50% of AC probands harbor mutations in one of them. Mutations in non-desmosomal genes have been also described in a minority of AC patients, predisposing to the same or an overlapping disease phenotype.

Compound/digenic heterozygosity was identified in up to 25% of AC-causing desmosomal gene mutation carriers, in part explaining the phenotypic variability. Abnormal trafficking of intercellular proteins to the intercalated discs of cardiomyocytes and Wnt/beta catenin and Hippo signaling pathways have been implicated in disease pathogenesis. The acquired and progressive myocyte death (either apoptosis or necrosis) typically starts in the subepicardial-midmural layers to move towards the endocardium. AC is a major cause of sudden death in the young and in athletes. The clinical picture may include a sub-clinical phase; an overt electrical disorder; and right ventricular or biventricular pump failure. Ventricular fibrillation can occur at any stage. Genotype-phenotype correlation studies led to identify biventricular and dominant left ventricular variants, thus supporting the use of the broader term...